Agenesis of the Corpus Callosum

Patient Information Series – What you should know, what you should ask.

What is Agenesis of the Corpus Callosum?

Agenesis of the Corpus Callosum (ACC) is a rare neurological condition where the corpus callosum, the major structure consisting of nerve fibers that connects the two hemispheres of the brain, is either partially or completely absent. This condition can be partial or complete and can occur as an isolated condition or as part of a syndrome with other abnormalities. Because of the timing of the development of the corpus callosum during gestation, agenesis of the corpus callosum cannot be diagnosed before about the middle of pregnancy.

The corpus callosum plays a crucial role in transmitting neural signals between the two hemispheres of the brain, facilitating communication and coordination. When agenesis of the corpus callosum occurs, it affects this interhemispheric transfer of information, which can lead to a range of outcomes.

What causes Agenesis of the Corpus Callosum?

The exact cause of ACC is often unknown, but it can be attributed to genetic abnormalities, prenatal infections or injuries, or exposure to harmful substances during pregnancy. In some cases, it is associated with chromosomal anomalies or inherited genetic conditions.

Should I have more tests done?

You might be referred for specialist neurosonography (ultrasound of the fetal brain and central nervous system) and magnetic resonance imaging (MRI). Further targeted ultrasound may be ordered to determine if other anomalies are present. Genetic counseling and genetic testing could be recommended. This may include tests like amniocentesis to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing. These specialists and your primary physician can help you make informed decisions about your pregnancy.

What are the things to watch for during the pregnancy?

You should have regular ultrasounds to assess the baby's well-being and development, and attend your regular prenatal care. Your caregiver will advise you as your pregnancy progresses, for example, pay attention to your baby's movements. Significant changes in the pattern or frequency might need to be evaluated.

What does it mean for my baby after it is born?

ACC on a prenatal scan does not predict the exact outcome for your baby. Some individuals with ACC lead healthy, typical lives, while others may a range of symptoms such as developmental delays, intellectual disabilities, and physical coordination issues, learning disabilities, difficulty in coordinating movements, social and behavioral challenges, seizures, or vision and hearing impairments. You might be referred to a pediatric neurologist or a specialist in fetal medicine for more detailed advice and to prepare for any necessary care after birth. You may be able to start planning for early intervention services. Even if your child shows no immediate symptoms, early involvement of specialists can be beneficial. The team caring for you and your baby during pregnancy can advise you how best to plan for delivery and afterward.

Will it happen again?

Each pregnancy is different, and the likelihood of a recurrence of ACC can vary greatly for different families, largely depending on the underlying cause. Genetic counseling can provide more personalized risk assessment and information. ACC can occur as an isolated condition or as part of various genetic syndromes or chromosomal abnormalities. When ACC is isolated the recurrence risk for future pregnancies is generally considered to be in the range of 1% to 2%, but this can vary. If ACC is associated with a genetic syndrome, the recurrence risk depends on the inheritance pattern of that syndrome. Determining the specific cause of ACC in an individual or within a family is crucial for providing an accurate recurrence risk.

What other questions should I ask?

- What are the long-term implications of ACC for my child's health, development, and quality of life?
- Are there any additional tests or evaluations that need to be performed during the pregnancy to assess my baby's condition further?
- What are my options regarding continuing the pregnancy?
- Where should I deliver my baby?
- Can you tell me about available treatments for my baby after birth?
- Can you provide information about the medical team and specialists who will be involved in my delivery and the baby's care after birth?

Last updated 4-2024